aminoacylase 1 deficiency

Aminoacylase 1 deficiency is an inherited disorder that can cause neurological problems; the pattern and severity of signs and symptoms vary widely among affected individuals. Individuals with this condition typically have delayed development of mental and motor skills (psychomotor delay). They can have movement problems, reduced muscle tone (hypotonia), mild intellectual disability, and seizures. However, some people with aminoacylase 1 deficiency have no health problems related to the condition. A key feature common to all people with aminoacylase 1 deficiency is high levels of modified protein building blocks (amino acids), called *N*-acetylated amino acids, in the urine.

Frequency

The prevalence of aminoacylase 1 deficiency is unknown.

Genetic Changes

Aminoacylase 1 deficiency is caused by mutations in the *ACY1* gene. This gene provides instructions for making an enzyme called aminoacylase 1, which is involved in the breakdown of proteins when they are no longer needed. Many proteins in the body have an acetyl group attached to one end. This modification, called *N*-acetylation, helps protect and stabilize the protein. Aminoacylase 1 performs the final step in the breakdown of these proteins by removing the acetyl group from certain amino acids. The amino acids can then be recycled and used to build other proteins.

Mutations in the *ACY1* gene lead to an aminoacylase 1 enzyme with little or no function. Without this enzyme's function, acetyl groups are not efficiently removed from a subset of amino acids during the breakdown of proteins. The excess *N*-acetylated amino acids are released from the body in urine. It is not known how a reduction of aminoacylase 1 function leads to neurological problems in people with aminoacylase 1 deficiency.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- ACY1D
- deficiency of the aminoacylase-1 enzyme

Diagnosis & Management

These resources address the diagnosis or management of aminoacylase 1 deficiency:

- Genetic Testing Registry: Aminoacylase 1 deficiency
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1835922/

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Seizures
  https://medlineplus.gov/ency/article/003200.htm
- Health Topic: Amino Acid Metabolism Disorders
  https://medlineplus.gov/aminoacidmetabolismdisorders.html
- Health Topic: Developmental Disabilities
  https://medlineplus.gov/developmentaldisabilities.html
- Health Topic: Genetic Brain Disorders
  https://medlineplus.gov/geneticbraindisorders.html
- Health Topic: Seizures
  https://medlineplus.gov/seizures.html

Genetic and Rare Diseases Information Center

- Aminoacylase 1 deficiency
  https://rarediseases.info.nih.gov/diseases/9741/aminoacylase-1-deficiency

Educational Resources

- Boston Children's Hospital: Muscle Weakness (Hypotonia)
  http://www.childrenshospital.org/conditions-and-treatments/conditions/muscle-weakness-hypotonia
- Disease InfoSearch: Aminoacylase 1 Deficiency
  http://www.diseaseinfosearch.org/Aminoacylase+1+Deficiency/374
- MalaCards: aminoacylase 1 deficiency
  http://www.malacards.org/card/aminoacylase_1_deficiency
- Orphanet: Neurological conditions associated with aminoacylase 1 deficiency
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=137754

Patient Support and Advocacy Resources

- Children Living with Inherited Metabolic Disorders (CLIMB)
  http://www.climb.org.uk/
- The Arc: For People With Intellectual and Developmental Disabilities
  http://www.thearc.org/page.aspx?pid=2543

Genetic Testing Registry

- Aminoacylase 1 deficiency
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1835922/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28aminoacylase+1+deficiency%29+OR+%28aminoacylase+I+deficiency%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- AMINOACYLASE 1 DEFICIENCY
  http://omim.org/entry/609924

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Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/condition/aminoacylase-1-deficiency

Reviewed: May 2014
Published: February 14, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services